F.I.R.S.T.
Foundation for Ichthyosis and Related Skin Types

ICHTHYOSIS FOCUS

Vol. 19, No. 3 A Quarterly Journal for Friends of F.I.R.S.T. Summer 2000

Ichthyosis Awareness Week October 1–8, 2000

Ichthyosis Awareness Week this year is scheduled for October 1 - 8. All F.I.R.S.T. members are asked to focus their efforts during this week to promote awareness about ichthyosis and the foundation. As we all know, ichthyosis does not receive a lot of media attention and we need to change that. I urge all of you to take the time to write to your local newspapers and television stations and share your personal story about ichthyosis. The local media is always looking for human-interest stories to share with its audience and your story may be just the one! It's simple and easy. Just write your personal experience of living with the disease (be sure to mention F.I.R.S.T. so people can contact us if they have questions), send a photo and put it in the mail. In a few days, call the newspaper editor or television station to make sure they received your letter and inquire about their interest in running your story.

Another way to support the foundation during Ichthyosis Awareness Week is to conduct a simple fund raiser and invite your local media to the event. This can be a lot of fun and take little effort. There are many creative ways to raise funds for F.I.R.S.T. Here are some suggestions, but the best ideas can come from you! Think of something you enjoy doing and turn it into a fund raiser!

1. Collect and recycle aluminum cans.
2. Have a bake sale or garage sale.
3. Organize a roller-a-thon, dance-a-thon or bike-a-thon.
4. Place collection cans in your local stores along with your picture and info about ichthyosis.
5. For a $1.00 donation, have your local businesses display a colorful butterfly with the donor's name on it. For more information about the "Butterfly Fund Raiser," contact Tracie Pretak at 814.929.5970.

And, as always, the national office is there to help you along the way. Contact Jean, Lori or Kelly to share your thoughts so we can turn it into a fun and rewarding experience for you.

Just think about the impact we could make if all our members made contact with their local media and/or conducted a fund raiser. Here's some encouragement:

- The more funds raised for F.I.R.S.T.....the more we can do to help our members.
- History has proven that the F.I.R.S.T. office always receives inquiries after someone has a story published or aired.
- With the awareness you create, a person who has been affected with ichthyosis may hear about the foundation for the first time and realize there are others who share their disease. Even if we help that one person, we have succeeded.

Good luck!

Do you need some financial assistance to help with your skin care? See page 3 about how you can apply for real cash to help purchase products and treatments for your ichthyosis.

2000 National Conference Wrap-Up

This year's family conference, F.I.R.S.T. Puts Me First, was a huge success! The conference was held in Philadelphia last month (July 7-9) at the Doubletree Hotel. See pages 7, 8 & 9 for pictures and fun facts about this year's event.

Please Give to the United Way

It's simple…
just write in Foundation for Ichthyosis & Related Skin Types (F.I.R.S.T.) on the Donor Choice Option Form and your funds will be designated to F.I.R.S.T.!

A United Way
System Partner

Thanks to all who contribute through the United Way.....
your help goes a long way!

Correspondence Corner

Dear Jean:

Several months ago I called you to get more information about F.I.R.S.T. At that time I knew very little about the organization or its membership. I am an individual who was born and have lived my entire life with X-linked ichthyosis. You and F.I.R.S.T. helped me to contact other people who have this disease and helped me to find the Ichthyosis Registry, which is here in Seattle at the University of Washington.

I wish to inform you that thanks to you I am now a member of the registry. I believe that it will be a great benefit to everyone who is affected with this disease.

Since joining F.I.R.S.T. and the registry I have found myself becoming more involved with the social and medical issues surrounding the disease. I was concerned when I learned that one individual was dismissed from her employment because her employer felt that her physical appearance, which was caused by the disease, was not acceptable to the company. Many of us have experienced such treatment from people in society, and my family and I fear discriminatory treatment and genetic profiling from employers.

I wrote a letter to my district congressional representative about this issue. Her name is Shay Schual-Berke, the Washington State Representative from the 33rd District. Ms. Berke is a doctor, so I believe she has a great knowledge and understanding of these issues. She brought my concerns to several administrative agencies within the government and helped me contact a group, which can provide assistance in regards to such situations. I was very surprised when she paid me a personal visit at my home. Such attributes are rare in a political representative and I greatly appreciated her help.

I just wanted to write and say thank you for all of your help over the past several months. I have learned a great deal about what it is like to have ichthyosis, and to know that I am not the only one who suffers from this disease. Thanks again.

Paul Goetzinger
Des Moines, Washington

* To contact the National Registry, call 800-595-1265.

Dear F.I.R.S.T.:

My name is Clem Amore and I am 46 years old and I too have Lamellar Ichthyosis. I have had it since birth. Toni's story (published in the last newsletter, Spring 2000) was very similar to my own as a child attending school in the early sixties and seventies. I could relate to her feelings of rejection and longing for a mate and a family. I am single, never married. My family has always been very loving and supportive, especially my dear mother Sue. She has always been my champion and has always taken awesome care of me. She has given me a strong faith in God and personal relationship with Jesus Christ and has given me the confidence to do whatever I set my mind to.

I too have a problem with dealing with the heat as I do not sweat and I have had my hands and feet crack and bleed. I play the guitar (since I was 12) and I am interested in and have a knack with computers. I have some dear friends that accept me for who I am on the inside. They have been my friends for a long time; some from grammar school and some from high school. I have a job with the Federal Government which I have been blessed to have for the past 28 years.

My current treatment is 40 mg of Accutane every other day, petroleum jelly and Aquaphor and various creams and lotions.

I am not seeing a dermatologist at this time. My medication is being monitored by my primary care doctor. He monitors my blood levels every six months to make sure the medication is not adversely affecting me. I have had problems with my eyes. Two ulcers on my left eye have left me legally blind in that eye. My right eye has relatively good vision. I have had two surgeries on both eyes to correct drooping lower eyelids.

Thank you for your newsletter. Growing up in the sixties and seventies there wasn't much support. I was fortunate to live in a major city (Chicago, IL) and I saw dermatologists at the University of Illinois Hospital. I even participated in many studies on oral and topical medications to see if they would help my condition.

It's comforting to know that now there is support and I am not alone. I would like to join the National Registry but I am not seeing a dermatologist. My treatment is being overseen by my primary care physician.

I can be reached by email at clemam@writeme.com. My mother purchased a one-year subscription to Ichthyosis Focus for me so my name and address should be in your database.

I have already placed orders for some of the products listed in the newsletter. Thank you for the resource.

In God's Love,

Clem Amore

Apply Today! Assistance Available for Ichthyosis Products and/or Treatments

F.I.R.S.T. realizes that the fight against ichthyosis is not only a medical one but a financial one as well. As families of affected individuals maintain their daily routine of treatments, it can be complicated by the ongoing costs of medical supplies and procedures.

Thanks to the generosity of an anonymous donor, the foundation has established the **Jane Bukaty Membership Assistance Fund** to help alleviate some of the financial burden that may be facing some of our members. It's easy to apply for and you will receive real cash! Here's what you need to do:

- ✔ Submit a letter indicating your need for funding.
- ✔ State the amount of money you are requesting.
- ✔ Demonstrate the financial need for this product/treatment.
- ✔ Indicate what product or treatment you want the funding for (some examples are):
  - Creams and lotions
  - Ear or eye products
  - Scalp or hair products
  - Cooling products (vests, collars, mister, etc.)
  - Bath products

Awards will generally not exceed $100.00. Applications will be awarded two times per year as determined by the Support Network & Member Assistance Committee.

Email your request today to info@scalyskin.org or mail your letter to the attention of the Jane Bukaty Membership Assistance Fund at 650 N. Cannon Avenue, Suite 17, Lansdale, PA 19446. You will be contacted by a member of the Support Network & Member Assistance Committee if you have been awarded aid from this fund.

Donations to the Jane Bukaty Membership Assistance Fund are always appreciated and enable F.I.R.S.T. to make this fund available to more of our members.

ASK THE READERS: What Do You Do?

At the national office, we continually receive questions from our members about many different issues. We know that many of you have your own "tried-and-true" methods for skin care. We also know that many of you may have great answers to those difficult questions that are frequently asked by the general public.

We have developed **ASK THE READERS:** What Do You Do? to share this valuable information with others. Each issue will ask a question of our readers and we will print your answers. If you have any advice you would like to share, please take a moment and send in your comments.

How do you remove scales from your scalp?

1. We rotate with a few different shampoos every couple weeks. The shampoos we like are T-Sal Extra Strength, PNS Liquid (available through pharmacy) and Biojoba Shampoo by Joyco (available through beauty salon and most economical). Then, every two weeks we apply mineral oil to our son's hair, rubbing it in very well. He then sleeps with a cap overnight and in the morning we use a fine-toothed metal comb to pull up the loose scales and remove them. **Beth G.**

2. Our strategy involves using a compound of 1 part lime water, 1 part olive oil and 6% lactic acid. We have a prescription for this.

We put it in a squirt bottle and apply it to her scalp in the bathtub when her hair is wet. We leave it on all night and wash it out the next day. If we apply it this way 2 or 3 times a week, it will loosen the scale and it will be much easier to remove with a comb. If the head is sensitive, you can leave out the lactic acid and it won't sting at all. You want to be careful to not let it get on skin that is open or recently had scale removed. It will sting like any cream with lactic acid in it. **Tiffany K.**

3. I coat my daughter's scalp at night with Lubriderm lotion and wrap her head with a bandanna. In the morning she takes a bath and I wash her hair twice with either T-Sal or Healthy Scalp, both by Neutrogena. After the bath I brush out her hair and use a very fine comb (a dog's flea and tick comb to be exact) to lift and pull the scales out. **Lori F.**

4. We apply mineral oil with a cotton ball to the scaly parts on my daughter's scalp. We let the mineral oil sit on her scalp for about one hour. Then we wash her hair with regular shampoo (my daughter doesn't like the smell of medicine shampoos) and we pull the scales out of her hair with a fine toothed-comb. **Sue G.**

Here's our next question:

How do you answer "what is wrong with you or your child's skin?"

Send us your comments. We will print responses in the fall 2000 newsletter. Here's how to contact us:

E-mail: newsletter@scalyskin.org
Fax: 215.631.1413
Call: 800.545.3286
Write: 650 N. Cannon Avenue, Suite 17, Lansdale, PA 19446

These helpful hints are provided by our members. F.I.R.S.T. does not recommend or endorse specific products or treatments. The information presented here should not be construed as medical advice and does not replace the counsel of your physician. Individual variances in ichthyosis cases require the consultation of a physician. It is important to see your doctor before altering anything in your treatment.

Ichthyosis Focus

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The editor invites your correspondence. We welcome your comments, observations and suggestions. Please send your letters to Ichthyosis Focus at the address listed above.

Executive Director's Report

Dear Members & Friends of F.I.R.S.T.:

It is always a pleasure to write this article for the newsletter because there are so many great things happening at F.I.R.S.T. This issue is even more special to me since I have the opportunity to share my feelings about the family conference.

I have just completed my first year as executive director and we have made great strides in strengthening the foundation, both financially and in membership. During my first year, I was only able to meet a few of our local members. But last month, at the family conference, I met over 250 of our members who have been affected by this disease. What an experience!

I was awestruck with what I saw and felt at the conference. It was extremely heart-warming to see so many people gathered in one place to help each other cope with ichthyosis. In addition to the medical information from our doctors, our members were able to socialize with old and new friends and share their personal experiences. Some of you even felt comfortable enough to talk personally with me about your lives. I gained a clearer perspective of what it is like to have the disease and what it is like to have a relative or friend with ichthyosis.

I was most impressed with the attitude and disposition of the children. Some of the kids were more severely affected than others, but they were immediately accepted into the "circle of friends." For a short time, everyone was the same. The teens quickly found each other, pulling up chairs to talk. The younger children shared their talents with us on Friday and Saturday night at the microphone and played with each other throughout the weekend.

Before July, I thought I had an idea of what the conference meant to our members, but now I truly know how important it is. If you haven't had a chance to attend a conference, I encourage you to start planning now! We are beginning to work on the next conference, sometime in summer 2002. I'll keep you posted with the details and let you know where it will be as soon as possible.

Jean

F.I.R.S.T. Medical Advisory Board Meeting

Saturday, March 11, 2000, the Medical Advisory Board met for its annual meeting in San Francisco at the American Academy of Dermatology conference. Several members of the Board of Directors joined the advisory board to discuss the foundation and plan for its future. The next annual meeting is scheduled March 2001 in Washington, DC.

Seated, from left: Glenn Oclassen, Mary Williams, M.D., Leonard Milstone, M.D., Donna Rice, Gloria Graham, M.D. Second row, from left: Michael Dunleavy, John DiGiovanna, M.D., Peter Elias, M.D., Mary Spraker, M.D., Amy Nopper, M.D., Bari Cunningham, M.D., Ho Jin Kim, M.D., Joseph McGuire, M.D., Jean Pickford, Susan Bayliss Mallory, M.D. Third row, from left: Peyton Weary, M.D., Eugene VanScott, M.D., Laurence Miller, M.D., Neil Prose, M.D., Robert Silverman, M.D., Philip Fleckman, M.D., Ervin Epstein, M.D., Alan Moshell, M.D., and Geoff Hamill, R.N.

F.I.R.S.T. Needs a Copy Machine

The national office is desperately seeking a copy machine to perform basic office functions. Our 11-year-old copy machine has seen its final days and we are having difficulty in keeping up with our daily work. Please contact Jean Pickford at 800.545.3286 to share your ideas or make delivery arrangements. Thank you!

Volunteer Opportunities

Hi! My name is Lori Schreiber, and I have been working at F.I.R.S.T. as the part time Program Director since May. My background includes counseling, teaching, and program development. In addition to my employment with the foundation, I currently teach counseling and sociology courses at a local community college and have a private counseling practice.

I live in a suburb very close to the F.I.R.S.T. national office, with my girlfriend Linda, and my two cats, Jem and Reisling. Being first time homeowners keeps us very busy, as home improvements seem to be a constant necessity. Summer, my favorite season, is filled with trips to the pool or beach and as many deck parties as possible!

This summer has been particularly interesting to me as I was able to attend the 2000 Family Conference. It was a pleasure to meet so many wonderful people and to hear so many inspiring stories.
I really wish that I had the chance to get to know even more of our members, and can't wait until the next opportunity!

This brings me to the business portion of my message. My job at the foundation is to organize and promote our various programs, particularly the Support Network, now called the ISN (Ichthyosis Support Network). In addition, we are also hoping to sponsor some regional meetings, if finances and volunteers are available. Please be aware that there are many ways to get involved in the organization, and I hope many of you will contact me to express your interest.

Finally, thank you to all the members, board members, and my wonderful co-workers who have helped me to feel so welcomed in the last few months. The transition into a new position, especially since I am only here a few hours per week, can be overwhelming. But everyone's dedication and enthusiasm makes coming to work a real pleasure. I look forward to many positive interactions with F.I.R.S.T. members throughout the year.

Lori

Programs, Special Events and Projects

How can you get involved?
We welcome volunteers to help us, and here are some ideas:

- ❖ Ichthyosis Support Network
  - Support Network Coordinators
  - Peer Counselors
- ❖ Grassroots Fundraising
  - Garage sales, Bake sales, Fun Runs, Beef and Beers, Golf Tournaments, Bowl-a-thon, Jars on Store Counters, etc.
- ❖ Media Awareness
  - Articles to local newspapers, call-ins to local radio stations
- ❖ Conference Committees
  - 2002 Family Conference Committee
  - Regional Meetings Committee
- ❖ Newsletter
  - Submit articles, pictures, stories
- ❖ Donor Gifts
  - Matching Gifts
  - United Way donations
  - Combined Federal Campaign
  - Memorials
- ❖ Ichthyosis Awareness Week
  - Sponsor one of the ideas above, or come up with another idea of your own.

Remember the national office will offer assistance with any of these projects. Call us with your proposals, and GET STARTED TODAY

Volunteer Recognition

Volunteer of the Year Award – Presented to members whose leadership and heartfelt dedication to F.I.R.S.T. best exemplifies the spirit of giving.

1999

The Eastin Family, *Brea, CA*

2000

Matthew Vecera, *Teaneck, NJ*

Frances Bernsteil Memorial Award – Presented to members whose outstanding grassroots fund raising efforts help contribute to the financial strength of the foundation.

Tracie Pretak, *Wilcox, PA*

Distinguished Service Awards – Presented to members for their extraordinary contribution or service to F.I.R.S.T. by helping us grow in strength, membership or recognition

Michael Dunleavy, *Blue Bell, PA*

Matt Gray, *Elkhorn, NE*

Elise Johnson, *Lynnewood, WA*

Ho Jin Kim, M.D., *Philadelphia, PA*

Ryan Licursi, *Cinnaminson, NJ*

Michelle Petersen, *New York, NY*

Deb Vilas, *New York, NY*

Volunteer Recognition – F.I.R.S.T. wishes to thank the following members for sponsoring programs, fund raising activities or promoting awareness in their local communities.

Lynne & Harry Alba, *Worcester, PA*

Debra Bowie, *Memphis, TN*

Nancy Cota, *Desert Hot Springs, CA*

Lori Florian, *Boulder City, NV*

William & Diana Grady, *Palermo, ME*

Tiffany Karst, *Shawnee, KS*

Mr. & Mrs. Brian McDougall, *Ontario, Canada*

Nicole McMillian, *Philadelphia, PA*

Grace McMillian, *Philadelphia, PA*

Patti Metzger, *Conesus, NY*

Liza Santamina, *Santa Clara, CA*

Susan Suda, *Grafton, ND*

Conference Tee-Shirts

Special thanks to Monna Morton for donating her creative expertise in designing our Tee-shirts. F.I.R.S.T. greatly appreciates her help! For more information, contact Monna Morton Graphic Designs at 215.473.3963 or email prattdsgn@aol.com.

Tee-shirts are available and can be purchased at the national office for $12.00 plus $1.50 for shipping. F.I.R.S.T. accepts Mastercard, American Express and Visa. Call today while supplies last. They're going fast! Call 1-800-545-3286.

Lighten Up for Goodness Sake!

by Mary Lilley-Thompson

Some of us think that if we lighten up, we're not taking life seriously. Well, I say "Hogwash!" Lightening up has nothing to do with the seriousness of life. It has to do with whether or not we're taking OURSELVES too seriously. You know you're taking yourself too seriously when you can't laugh at yourself.

Some of us can't lighten up because we don't want to give up being crabby. We wallow in 'crabby'. Nothing is a crab's fault. People know us by the crab lines between our eyebrows.

Today, try laughing at yourself in a mirror for thirty seconds. I dare you to take yourself less seriously. If you do, you will radiate a light that will leave a trail of light so that others might dance, shine and celebrate life.

(Note: Mary was our memorable closing keynote speaker at the National Family Conference in Philadelphia. If you would like one of her "Crabby" hats ($25 each+S&H), you can call her, toll free, at (877) 837-7446, or e-mail her: crabbyhat@marystarshine.com. For each hat purchased, Mary has generously offered to donate $5 to the Foundation. If you'd like to see the hat, go to www.marystarshine.com. and click on the button for "Who is Mary Starshine™?")

The survey results are in...

23 new families attended their first family conference.

One member traveled as far as 3600 miles to attend this year.

On a scale of 1 to 10 (10 being the highest) the conference was rated 8.7.

Kiddie Corp, our professional child care program, was a big hit with parents and kids.

100% of participants would recommend the family conference to others.

Our biggest successes: organization and professionalism, access to knowledgeable doctors, welcoming atmosphere, opportunity to meet and share with others.

Areas that need improvement: need more activities for teenagers, smaller breakouts, more time to socialize, a longer conference, more funds to do it once a year.

A young participant in the child care program

Registration volunteers anxiously await the arrival of F.I.R.S.T. members

Ben Franklin, a very special guest, stops to chat with a young member

F.I.R.S.T. members register and receive their goodie bags

Thank you for your hard work and dedication to the foundation. The conference was wonderful. I look forward to many more.

Barbara R.

Patiently waiting for some refreshments on Friday night, a young guest smiles for our camera

Congratulations! What a great job you did! The conference was so well done and everyone I spoke to seemed very happy being there and feeling supported. I hope you were pleased with the results and received a lot of positive feedback.

Kathy G.

A star is born...

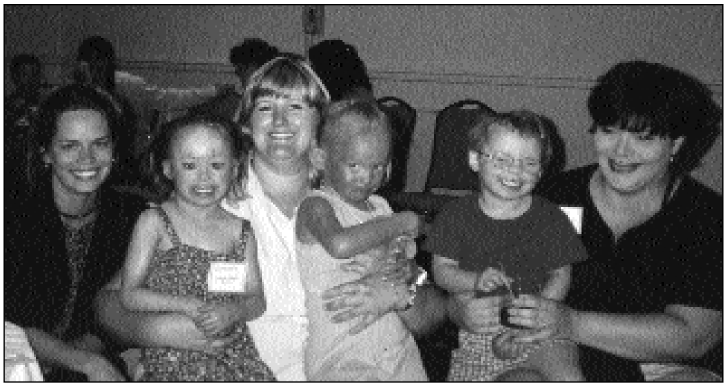
Everyone listens intently as someone shares their personal experience in the Mom's Discussion Group

I just wanted to let you know that I had a great time at the conference. It was the second one that I attended and I made some new friends and met many of the people that I talk to on-line.

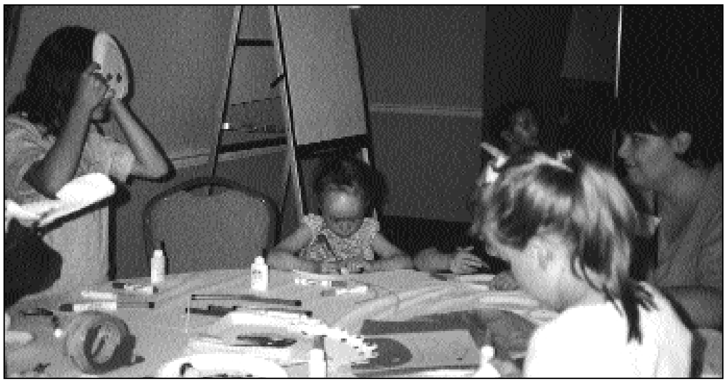
It was a great experience for my son too. He was able to see other kids who have skin like his. He really had a good time with Matt.

The weekend went by too quickly! Take care and thanks for organizing a great conference.

Denise B.



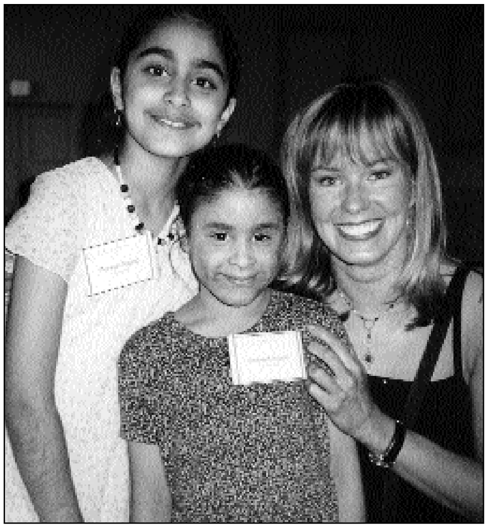
Smiles abound at Friday night's reception



Happy faces create silly masks

Playing games and making new friends in the Tween Breakout Group

Someone shares a funny story at the Women's Group Discussion



And the winners of the smiling contest are…

Thank you so much for everything you did at the conference. My family and I were very glad that we went. It was nice to meet you and the other members of F.I.R.S.T. We learned a lot more about ichthyosis. These past eight months have been an adjustment to the daily regime for our son, Timmy, but the foundation has helped tremendously.

Mary H.

The competition is fierce as these young girls play a game with one of our child care volunteers

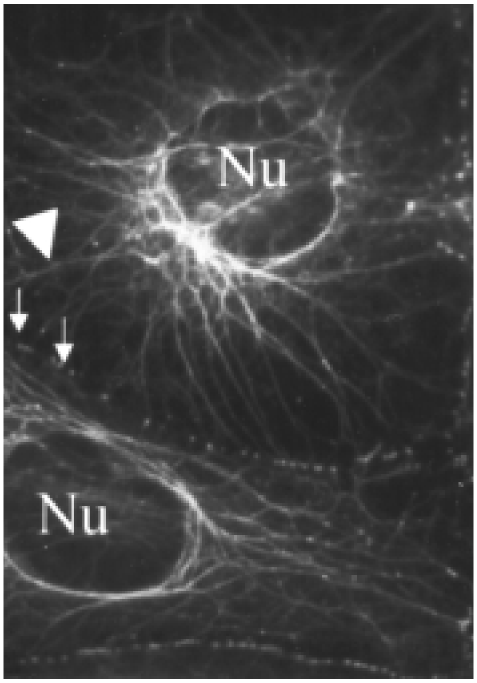
Guy talk... Dads and men come together to share with each other

Cell – Cell Coupling and Skin Diseases

Peter J. Koch, Ph.D.,
Departments of Dermatology & Molecular and Cellular Biology, Baylor College of Medicine, Houston, Texas

One of the fascinating aspects of skin research is the symbiosis that developed in recent years between scientists from different fields, such as molecular and cellular biology, biochemistry, and dermatology. The contributions of specialists from these fields have resulted in dramatic advances in our understanding of normal skin development as well as certain skin diseases.

My research has focused on the epidermis, the uppermost cell layers of the skin. For a biologist, it is of particular interest to understand the molecular mechanisms that regulate the life cycle of epidermal cells. The epidermis is a self-renewing tissue. Relatively few cells within this tissue have the capacity to divide indefinitely (‘stem cells’) and to develop into more specialized cells. This process, termed differentiation, results in the formation of the different cell layers that form the epidermis. The endpoint of this process is the formation of a layer of dead cells (‘stratum corneum’) that covers and protects the body surface from environmental hazards. Concomitantly with this differentiation process, epithelial cells migrate from the lower epidermis towards the body surface where they are eventually sloughed off from the stratum corneum. During differentiation, the epidermal cells have to maintain stable cell-cell connections to ensure that the epidermis remains intact. A cellular structure that is essential for this function is the desmosome. Desmosomes are disc-shaped subcellular protein complexes that not only couple neighboring cells, but also provide anchoring points for intermediate filaments (IF). IFs are part of the cytoskeleton which is essential for the mechanical stability of epidermal cells. Impaired desmosome function has been linked to skin diseases. In pemphigus (pemphigus foliaceus, PF; pemphigus vulgaris, PV), autoimmune diseases in which patients develop skin and mucous membrane blisters, autoantibodies against desmosomal proteins (desmogleins) were detected. It has been shown that these desmoglein antibodies, when injected into mice, can induce the symptoms of pemphigus. These experiments clearly established a causative link between these antibodies and pemphigus.



Microscopic picture of two epithelial cells that form desmosomes (seen as small dots demarcating the cell membranes, two desmosomes are marked by arrows) and intermediate filaments (IF, one filament labeled with arrowhead). The IFs of the two cells form networks that extend from the region around the cell nuclei (NU) towards the cell membranes where they bind to desmosomes. Desmosomes appear as ‘pearls on a string’.

Pathogenic antibodies in PF are binding to desmoglein 1 whereas pathogenic antibodies in PV mainly bind to desmoglein 3. Both desmogleins are transmembrane proteins; i.e. they are embedded in the cell membrane and have extra- and intracellular domains. The extracellular domain is thought to contribute to cell-cell coupling.

To further define the biological function of a desmoglein in a living organism, we recently generated desmoglein 3 knockout mice, i.e. mice in which the gene for desmoglein 3 was switched off. This mouse model recapitulates some of the features of PV and thus supports the view that the symptoms of PV are caused by a loss of functional desmoglein 3.

Desmosomal proteins are not only affected in autoimmune diseases. Recently, mutations in desmosomal genes were linked to a form of palmoplantar keratoderma, a thickening of skin on palms and soles, (desmoglein 1 and desmoplakin genes) and ectodermal dysplasia/skin fragility syndrome (plakophilin 1 gene). Taken together, the above mentioned results clearly indicate that intact desmosomes are crucial for the normal development and function of the skin.

There is another class of desmosomal proteins that I have not mentioned so far, the desmocollins. Like the desmogleins, desmocollins are transmembrane proteins. Cell culture experiments from several laboratories indicated that these proteins might be essential for desmosome-mediated cell adhesion as well as connecting the desmosomes to the IF cytoskeleton. However, little is known about the role of these proteins in desmosome-function in skin. Recently, it has been reported that the sera of patients that suffer from a subtype of ‘IgA pemphigus’ contain anti-desmocollin 1 antibodies. It has been suggested that these antibodies might contribute to the disease symptoms. However, experimental evidence to support this hypothesis has not yet been published. Nevertheless, based on previous biochemical and cell culture experiments, one has to assume that impaired desmocollin 1 function would lead to severe skin disease. One way to test this hypothesis is to introduce mutations into the desmocollin 1 gene of mice and study the effect of these mutations on the skin. Several types of mutations can be tested. A classical approach is to switch off the gene, i.e. to generate a ‘knockout’ mouse. The analysis of knockout mice allows us to draw conclusions towards the normal function of the protein. Another approach is to introduce more subtle mutations into the desmocollin 1 gene that affect only certain domains of the protein. This approach allows an assessment of the role of individual protein domains in cell-cell coupling and IF anchorage.

Besides new insights into the biological function of desmocollins, we can expect clues from these mice with respect to human diseases that might be caused by desmocollin mutations. Those clues will enable us to define symptoms caused by desmocollin mutations and to specifically test patients with similar disease symptoms for desmocollin mutations. The next step will then be to use these animal models to develop new treatments for these diseases.

Acknowledgments

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Volunteers Needed for Research Study

Nicholas J. Lowe, M.D., Clinical Professor of Dermatology at the UCLA School of Medicine is currently enrolling patients with ichthyosis, Darier's Disease and related disorders of cornification for a study to evaluate the efficacy and safety of tazorac 0.1% gel compared with Lac-hydrin 12% cream. All patients will be treated with either Tazarotene gel or Lac-hydrin cream and your responses will be evaluated. Treatment and medical visits will be free for patients and you will be paid $50.00 per visit, approximately $400.00 upon completion of the study. Please contact Teresa Bourget or Dary at Clinical Research Specialists at 310.828.8887.

1st Annual Trick or Treat Fun Run in Friendswood, TX

The 1st Annual Trick or Treat Fun Run, organized by Dawn Johnson will be held on October 14, 2000 in Friendswood, TX at 8:00 a.m. For more information about how you can get involved, please call Dawn at 281.996.8902.

The Magic of Mermaids

by Shelly Menzia


Illustration by Merritt Andrews

Do you have dreams that are so real they haunt you for the rest of your life?

There is one in particular that I remember when I was about eleven years old. I was out camping with my family near the Copper River in Alaska in an old beat up camper, on an even older powder blue pickup truck. In my dream a “merman” came to the camper to take me from my family. He was not like the beautiful mermaids or mermen from the movies or books. Although human like in form he had webbed feet and hands and huge “Spock” like ears. Prince Charming he was not. He was tall with various shades of dark, seaweed green skin. He told me I had to go with him and marry him under the sea. I was terribly upset that my family did not try to stop him. It was like they had been expecting it. My family shrugged their shoulders, looked at me sadly and waved good-bye as he pulled me away. I waved back crying and begging for him to let me stay with them. He quietly but firmly pulled me along.

When we entered the water, I was amazed that the water felt so warm and comfortable and that I could breathe. I had this amazing sense of freedom as we swam gracefully through the dark sea. In the distance from up above I could see the sun shining and sparkling down towards me and I was no longer afraid of the merman. After that I woke up but the dream has stayed with me for thirty years.

It wasn’t until many years later I was able to analyze that dream with a friend and understood that it actually had some meaning to it. You see I was born with a rare congenital birth defect called ichthyosis (fish skin).

It made growing up in our “soft skin is beautiful society” more difficult yet at the same time it shaped me into who I am. My skin is very dry with little elasticity, it has a “scaly” like appearance which sheds constantly.

The only place and time I am truly comfortable is when I am in the water. I spend hours in the tub and shower and put lotions on constantly so that I can feel as comfortable out of the water as I do in the water.

I have faced some discrimination with it, not being allowed into a public pool; when I was a kid, having people being afraid to touch me for fear it was contagious and having my blood be refused when I tried to donate it at a blood drive at college. These were just a few of the things that happened along the way.

I think the worst and most devastating incident however, was having my supervising teacher during my student teaching assignment ask that I not work with her anymore. She felt my skin was too “distracting” for the students to learn. I was devastated and I came very close to giving up my career in special education. I wanted to lock myself away in a room away from other people because of her prejudice and inability to understand.

Afterwards, I went home and had a good cry, (and consequently, had the rest of my family crying) and went to take a long bath. (So long in fact my mother came to check on me,. I think she had thought I had drowned myself or something.) I got through it though, and was placed with another woman who was a wonderful teacher and she was also very understanding. She knew what it was like to have a skin problem, she had skin cancer. She, along with my friends and family helped me get back on my feet emotionally and helped build up my confidence again. I ended up being much stronger for it and much less likely to place my beliefs about myself on the basis of one other person’s opinion. Especially someone I hardly knew.

Even though I was stronger however, my skin was still an issue with me. Oh, sure I could “handle” it OK and in 13 years of teaching I have never had a problem related to it . Still, my perception of myself was fairly low as far as physical attractiveness goes. I didn’t see myself as the person first, who happened to have ichthyosis. I was the girl with fish skin and somewhere inside of her was me.

Then something strange happened. As I was talking about my skin condition to a new friend, I was joking, “Yeah, I’m part fish and part human...... Hey, I guess that makes me a mermaid!” In uttering that one goofy sentence, I went straight from monster girl with fish skin to a beautiful mermaid swimming and frolicking under the sea. My perception of myself was changed radically and instantaneously from then on. You have heard of “black pride”, “deaf pride” and “gay pride”, with me it has became “mermaid pride”. The many long baths I had to take, once tedious and boring, became trips to the sea in my imagination or I was like Daryl Hannah in the movie Splash. Just going in to wet my “fins”. It was a magical transformation.

After that I started doing some research and became fascinated with the legends of mermaids. I learned that mermaid myths exist in almost every culture and they were thought by some to be healers or to have psychic powers. There have been very credible people who have sworn that they have seen mermaids, sometimes right before a storm as if they were being warned of danger. Many paintings show mermaids singing on top of rocks in the surf or playing with fellow sea creatures. Most of them are beautiful romantic images.

Then I started developing my own theory about where the legends of mermaids may have come from. What if just maybe, the legends of mermaids really did start with people who had ichthyosis? I had to have inherited it from ancestors up to ten generations back and it had to come from both sides of the family, (recessive genes); Irish from my dad’s side and English/ Dutch from my mother’s, both cultures which have mermaid myths.

Years ago people didn’t have lotions and baths to make them more comfortable so I imagine that everyone who had the chance would spend a lot of time in the water such as lakes, rivers or oceans trying to stay comfortable. Perhaps people saw ichthyosis people in the water and along with seeing their fish like skin, the legends of mermaids were “spawned”.

(Ichthyosis although rare is found in most cultures just like the legends of mermaids.)

Then I remembered going to an ichthyosis conference where for the first time I met other people with ichthyosis. I was amazed as I got to know people, of the common similarities we had besides our skin. We talked about some psychic experiences most

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> “In general, the goal in taking care of ichthyosis is to hydrate (moisturize) the skin, hold in the moisture, and keep scale thickness to a minimum.”*

**Foundation for Ichthyosis & Related Skin Types, http://www.scalyskin.org*

Aquaphor® Healing Ointment helps heal dry skin associated with ichthyosis. Its unique petrolatum-based formulation combines a moist environment with the benefits of a semi-occlusive barrier that allows skin to breathe and absorb fluids.

Aquaphor is ideal for daily use because it is hypoallergenic, non-comedogenic, fragrance and preservative-free. Aquaphor Healing Ointment is safe enough for even the most sensitive skin.

Special Offer

For Readers of this F.I.R.S.T. Newsletter

MANUFACTURER’S COUPON | EXPIRES 12/31/01

Save $2.00
on any
Aquaphor®
Product

(NO TRIAL SIZES)

Consumer: Coupon good on the purchase of any Aquaphor® Product, no trial sizes. Limit one coupon per item purchased. Any other use constitutes fraud. Not good with any other special offer. Consumer must pay sales tax.

Retailer: You are authorized to act as our agent to redeem this coupon and we shall reimburse you at face value plus 8¢ handling in accordance with our redemption policy. Cash value 1/100¢. Copies available upon request. Offer void if copied and where prohibited, taxed or otherwise restricted. Mail to: Beiersdorf Inc, PO Box 880504, El Paso, TX 88588-0504. Good only in USA.

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The Magic of Mermaids

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of us have had and I found out that almost every single person was musically inclined. Many were involved in choirs.

So of course, now I picture all our ancestors being the ones singing on those rocks and warning past sailors of danger.

Since then I have acquired quite the mermaid collection. Of course people who don't know me have a hard time figuring out why I have all these mermaids many which are bare chested mermaids all over the house. (I have thought about making them little bras so the more sensitive people won't be offended .) My friends have been wonderful and always keep their eyes open for special mermaid "treasures" when they are traveling. My most recent addition came from Italy.

I have had some fun with my mermaid revelation. Once in the Galapagos Islands I was on a Zodiac boat with some of the male guides from the ship.

They asked me about my skin but I didn't think they would understand my English and I knew I wouldn't be able to explain it in Spanish so I simply smiled at them and said very matter of factly, "Yo soy la sirena" (I'm a mermaid) and then I promptly dove into the water. When I looked back at them they had their mouths hanging open as they looked back and forth between me and each other. They couldn't decide if I was crazy, joking or maybe somehow telling the truth. They looked at me strangely throughout the rest of the trip. It was all I could do to keep from cracking up laughing every time they gave me one of those "looks". By the end of the trip I had been invited by the guides to sing and play with their on board music group they had formed. We had a blast. It was a fun and magical trip. My computer log-on name is now "Sirena".

It was just a few years ago however, that I was discussing the dreams I've had with my friend Lydia. When I told her about my merman dream she pointed out quite easily what it meant. The merman was the ichthyosis and I was married to it. I didn't like it, my family didn't like it, but there was nothing any of us could do about it. It suddenly seemed so simple.

Now, since I had been swimming through the "sea of life" for some time I realize it's OK to have ichthyosis and to be who I am. It's not any scarier or worse than I choose to make it. Whenever I want to I can swim up to that sun shining and sparkling down towards me from the surface to be who ever I want to be or do what ever I want to do, because that is where the magic lives. The only thing that can ever stop me from going anywhere or doing anything is my perception of myself and all I can say about that right now is......Mermaid Power!!!

What's New… What's Hot

Product for Redness

I have rosacea, a redness on my left cheek and nose. I found "Chang Sheng Herbal Beauty Soap" from the mail order catalog, **Lifestyle Fascination**, in Lakewood, NJ 1-800-669-0987. It has been a godsend and almost completely removed the redness. The texture of my skin is so much better than it ever was. I thought maybe this could help others who suffer with redness on their skin.
Mrs. H.McA, Plymouth Meeting, PA

Cream Available

If anyone uses Calmurid Cream or would like to try it I have several tubes to give away free of charge. The active ingredients are urea and lactic acid. Call Cindy Bremmer at 503.397.3082 or write to 834 Cedaroak Street, St. Helens OR 97051.

Occlusion Suit Available

A young girl's occlusion suit (size 10-12) has been donated to the office and is available to a member of F.I.R.S.T. on a first-come first-serve basis. Please call the national office at 800.545.3286.

What Worked for Me

I have found something to greatly help my Ichthyosis. I have a type that affects only my feet (mostly) and hands. I saw an article from Reuters Health describing how Fatty Acids and antioxidants appear to be an effective treatment for inflammatory injuries also known as 'tennis elbow' and 'golf elbow'. I didn't know about those two, but I knew my right knee hurt and was getting harder to walk with it.

I decided to try this treatment using a product at the local vitamin store called "Ultra Oils" made by Country Life of New York. It had the ingredients that the article called for which are as follows for each softgel: alpha-Linolenic Acid (Omega 3) 350 mg; Linoleic Acid (Omega 6) 300 mg; Oleic Acid (Omega 9) 250 mg; Gamma-Linolenic Acid 20 mg.

On May 1st I started taking 2 softgels with lots of Vitamin C and E each day. After 2 weeks I started noticing my knee getting much better and after 3 weeks it was really good but also noticed something was going on with my feet.

Now after 7 weeks (I upped the softgels to 3 a day on the 4th week) what knee pain and my Ichthyosis is much, much better. Yes, I still have it but the skin is not as red, its much softer and no "cuts" in the skin; I could not be happier. Also the skin appears to still be getting better as I proceed with this "treatment".

I do not know whether anyone else has tried these Fatty Acids for treatment and the result they got but I highly recommend others try and report on the results. You must also take Vitamin C (I take about 6,000 mg a day) and Vitamin E (2,000 mg a day) with the Fatty Acids.

Your organization is doing a great job and I am proud to be a member.
Jack E. King, Glendale, CA
kingcpa@earthlink.net

Past Newsletters

Visit our members only site at www.scalyskin.org. Past issues of the Focus Newsletter are available to all our members. Call the national office at 800-545-3286 to receive your password and user ID number to log on.

URGENT: MEMBER PRIORITIES SURVEY!

Shape the Future of F.I.R.S.T. By Speaking Up!

How are we doing so far? Where should we focus our efforts in the future? Your responses to this survey will help set our priorities and shape our programs as we approach our 20th Anniversary.

- ✰ Please feel free to duplicate this form so each family member may respond. (Parents, siblings, etc.)
- ✰ Completed forms should be mailed by OCTOBER 15, 2000 to:

  F.I.R.S.T. Member Surveys
  650 N. Cannon Avenue, Suite 17
  Lansdale, PA 19446
- ✰ This form is also available at our website www.scalyskin.org and can be filled-in and filed electronically.

1 How did you find out about F.I.R.S.T.?

Medical Professional Referral
Other F.I.R.S.T. Member
Other Support Organization
Literature Search
Internet Search
Other____________________________

2 What are our strengths and weaknesses? Please indicate your level of satisfaction with each item listed below.

	not satisfied	somewhat satisfied	satisfied	very satisfied	no opinion
Ichthyosis Focus Newsletter	☐	☐	☐	☐	☐
National Conference	☐	☐	☐	☐	☐
Ichthyosis Support Network	☐	☐	☐	☐	☐
F.I.R.S.T. Website	☐	☐	☐	☐	☐
Publications	☐	☐	☐	☐	☐
Responsiveness of Office Staff	☐	☐	☐	☐	☐
Physician Referrals	☐	☐	☐	☐	☐
F.I.R.S.T. Support of Research	☐	☐	☐	☐	☐

3 How does your overall experience with F.I.R.S.T. compare with your original expectations?

☐ much worse ☐ worse ☐ as expected ☐ better ☐ much better

4 Which three (3) areas are most important in terms of the future direction of F.I.R.S.T.? Please rank by placing 1, 2, and 3 next to your selections, with 1 being the most important (choose only three).

☐ Member Support Network
☐ Increasing Public Awareness
☐ Funding Research Grants
☐ Booklets and brochures
☐ Advocating for More Research
☐ Ichthyosis Registry
☐ Financial Assistance Program
☐ Other: ____
☐ Web Site Enhancement
☐ Ichthyosis Focus Newsletter
☐ Conferences

5 Which three (3) areas are least important in terms of the future direction of F.I.R.S.T.? Please rank by placing 1, 2, and 3 next to your selections, with 1 being the least important (choose only three).

☐ Member Support Network
☐ Increasing Public Awareness
☐ Funding Research Grants
☐ Booklets and brochures
☐ Advocating for More Research
☐ Ichthyosis Registry
☐ Financial Assistance Program
☐ Other: ____
☐ Web Site Enhancement
☐ Ichthyosis Focus Newsletter
☐ Conferences

6 Any Comments? (use an additional page if necessary): ____

____

7. Tell us about yourself:

☐ I have ichthyosis
☐ I am a medical professional
Your age:
Your sex: ☐ Male ☐ Female
☐ A family member/friend has ichthyosis: Relationship: ____________ Age: ______
☐ Other:____
Have you attended a FIRST conference? ☐ Yes ☐ No
Have you logged on to F.I.R.S.T.'s web site? ☐ Yes ☐ No

News & Notes

National Institute of Health's Website

Nielsen/Netratings, a subsidiary of ACNielsen, the world's leading supplier of market research information, has published new data showing that the NIH's web site has the largest audience of any government web location, including all federal, state and local sites. You can access the NIH's web site at www.nih.gov.

A recent new addition to the NIH web site is the Office of Disease Prevention location. The site, http://odp.od.nih.gov/ includes a collection of resources for patients and the public, including access to the NIH Rare Diseases Clinical Research Database. The page contains a listing of upcoming workshops and conferences on disease prevention and public health. Access to the "Population-Based Prevention Research Database" will be added soon.

Tax Credit for Families Attending the Medical Conferences

On May 8, 2000, the Internal Revenue Service (IRS) issued a ruling that will allow parents of children with chronic illnesses to deduct some of the costs associated with attending medical conferences related to their children's condition. The rule allows parents to deduct the expenses of admission and transportation to a medical conference related to the chronic illness of the individual's dependent. It is explained in Internal Revenue Bulletin 2000-19, which is located on the IRS website at http://www.irs.gov/bus info/bullet.html.

Excerpt taken from Alliance Alert, May/June 2000, A publication of the the Genetic Alliance.

Completion of Draft Human Genome Sequencing

Government scientists, industry researchers and consumer advocates joined President Clinton at the White House on June 26 to witness a joint announcement on completion of the draft human genome sequence. Francis Collins, Director of the National Human Genome Research Institute and Craig Venter, President of Celera Genomics, gave powerful speeches on the significance of this scientific achievement and the hope it brings for the future of medicine and public health. President Clinton advocated for rapid completion of necessary ethical, legal and social protections to ensure the full benefits and promise of genetics research. At a press conference later in the day, spokespersons for the parallel efforts emphasized the importance of the next research challenges and called for continued public support to build on these momentous scientific milestones.

Excerpt taken from Alliance Alert, May/June 2000, A publication of the the Genetic Alliance.

Please help us to minimize our printing costs. Call us at 1-800-545-3286 if you do not wish to receive any future issues of *Ichthyosis Focus.* **Help F.I.R.S.T. reduce its postage costs – would you be willing to accept the Focus Newsletter via e-mail? Let us know**

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**URGENT:
Member
Priorities
Survey
Inside**